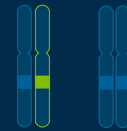
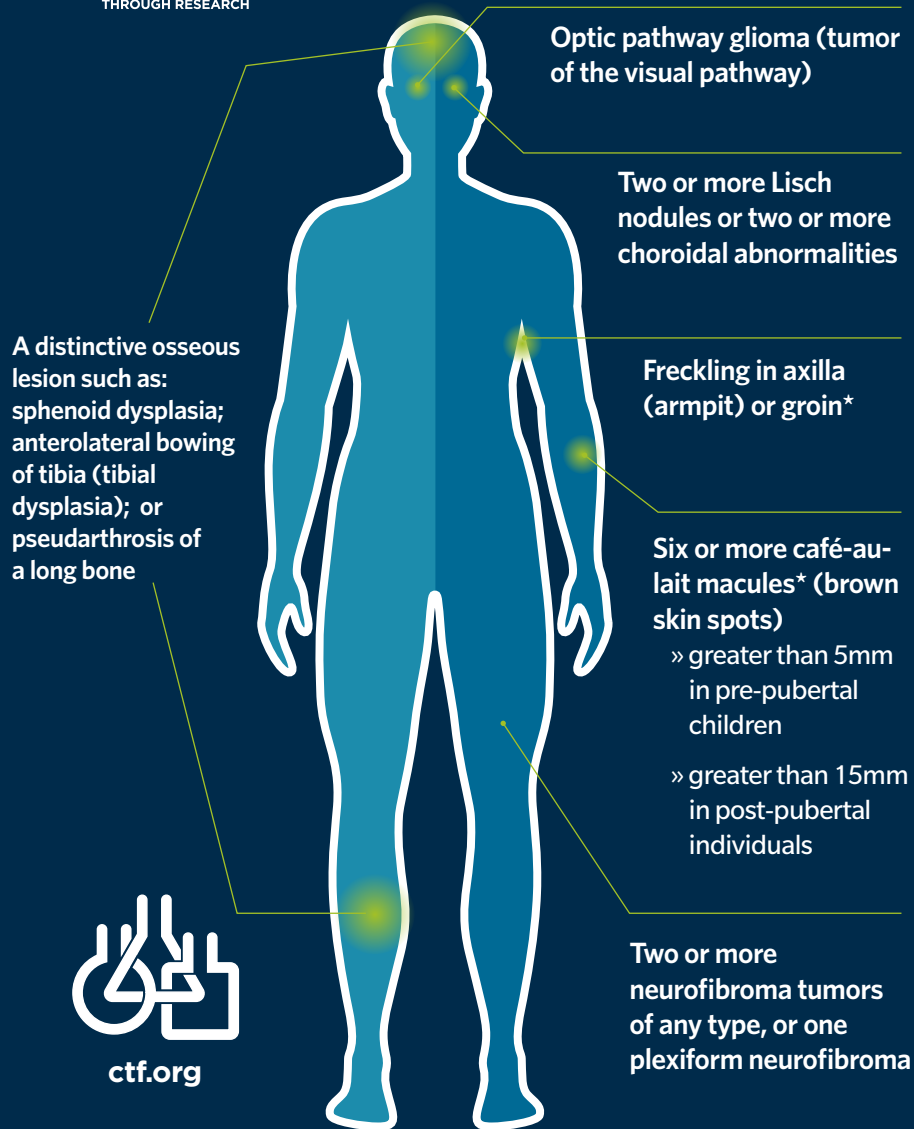


# DIAGNOSTIC CRITERIA FOR NEUROFIBROMATOSIS TYPE 1 (NF1)

2021 UPDATE

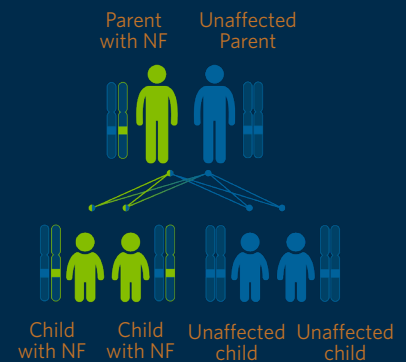
A diagnosis of NF1 can be given if an individual has two or more of the following manifestations.



## A pathogenic *NF1* gene variant\*\*

\*\*Additional Genetic Criteria Updates:

- The term “mutation” is no longer accepted; pathogenic variant is now the preferred term.
- Genetic analysis is not REQUIRED for diagnosis but may allow for an earlier diagnosis.
- Genetic analysis ALONE is not sufficient to diagnose NF1 - diagnosis requires a second diagnostic feature of NF1.



## A parent with NF1 based on diagnostic criteria

\*At least one of the two pigmentary findings (café-au-lait macules or freckling) should be bilateral.

## GLOSSARY

**Anterolateral bowing of tibia** is a curving of a bone in the leg

**Café-au-lait macules**, also called café-au-lait spots, are flat darkened areas on the skin

**Choroidal abnormalities** are problems with the vascular layer of the eye (the choroid)

**Dysplasia** means abnormal growth

**Lisch nodules** are small tan or brown bumps on the surface of the iris in the eye

**Neurofibroma** is a tumor that forms on a nerve cell sheath

**Osseous lesion** is a problem with a bone that is not due to injury

**Optic pathway glioma** is a tumor of the visual pathway

**Pseudarthrosis** is a condition that occurs when the bones in the leg do not heal correctly

**Sphenoid** is a bone near the base of the cranium behind the eye

A link to the NF1 diagnostic criteria update publication can be found at: [ctf.org/criteria](https://www.ctf.org/criteria)

Learn more about all types of NF on the Children's Tumor Foundation website at: [ctf.org](https://www.ctf.org)

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